Sea-Blue Histiocytosis in the Small Bowel

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Abstract

Sea-blue histiocytosis accompanies a variety of conditions including genetic and acquired lipid metabolic or ceroid storage disorders (such as type B Niemann–Pick disease), myelodysplastic disease, thalassemia, and secondary infections. A case of sea-blue histiocytosis is demonstrated with an involvement of the entire small bowel, probably caused by kalaazar. In the video, double-balloon endoscopy demonstrates the involvement of the small bowel. This article is part of an expert video encyclopedia.

Keywords

Double-balloon endoscopy; Enterscopy; Histiocytes; Small bowel; Video.

Video Related to this Article

Video available to view or download at doi:10.1016/S2212-0971(13)70119-5

Technique

Double-balloon endoscopy (DBE).

Material

Enteroscope: Fujinon EN-450P5, Fujifilm, Japan.

Case Report and Endoscopic Procedure

A 23-year-old woman presented with acute abdominal pain in the left flank and hepatosplenomegaly, associated with sporadic vomiting episodes and fever for 2 months. She had a past history of visceral leishmaniasis (kala azar, diagnosed 11 years before). Laboratory examination showed hemoglobin of 11.6 g dl^{-1}, leukocytes of 3.68×10^3 µl with a normal differential, and platelets of 309×10^3 µl. Liver biochemical test results were normal.

Bone marrow aspiration demonstrated the presence of numerous macrophages, suggestive of sea-blue histiocytosis. Forms of *Leishmania* were not observed.

Computed tomography scan revealed periaortic and mesenteric lymphadenopathy with a maximum diameter of 3.1 mm.

Upper gastrointestinal endoscopy revealed multiple sessile elevated and confluent lesions at the duodenum. DBE by the oral route not only confirmed numerous sessile lesions but also pedunculated and multilobated lesions that formed bridges throughout the lumen of the small intestine. DBE by the anal route revealed sessile ileal lesions enhanced by chromoscopy with indigo carmine. The esophagus, stomach, and colon were normal.

Biopsies were performed and May–Giemsa staining identified subepithelial sea-blue macrophages.

Background

Sea-blue histiocytosis is a disorder associated with both acquired conditions of increased cellular turnover and inborn errors of lipid metabolism.[1,2]

Silverstein *et al*. in 1970 first described sea-blue histiocytosis as a clinical entity.[3]

Clinically, it manifests with impaired liver and lung function, hepatosplenomegaly, and lymphadenopathy.

Diagnosis can be performed from the first year of age to the eighties but is usually made by the fourth decade. Approximately 15% of cases progress to cirrhosis and portal hypertension. Similar incidence is observed in men and women.[4]

The common feature is the accumulation of unsaturated lipids in various organs with lipid-laden histiocytes due to increased production or a failure of catabolism. Blood disorders such as chronic myeloid leukemia, idiopathic thrombocytopenic purpura, and myelodysplastic syndromes, in which cells are at an increased rate by the reticuloendothelial system, represents the former group. A variety of inherited metabolic defects including sphingomyelinase deficiency such as Niemann–Pick disease, Gaucher disease, and abnormalities of lipoprotein metabolism, characterize the latter group.[1]

Niemann–Pick disease is a congenital (inherited) autosomal recessive metabolic disorder caused by a deficiency of the enzyme acid sphingomyelinase coded by *SMPD1* gene. In contrast with type A Niemann–Pick disease, a severe neurodegenerative disease of infancy, type B Niemann–Pick disease patients have little or no neurodegeneration, and frequently

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survive into adulthood. Measurement of acidic sphingomyelinase enzymatic activity in peripheral blood leukocytes is helpful for the differential diagnosis of the various lipid metabolic disorders and ceroid storage diseases.[2,5,6,7]

In patients with myelodysplastic syndromes, the accumulation of histiocytes in the bone marrow is probably associated with excessive turnover of hematopoietic cells. Under these circumstances, macrophages may be unable to metabolize completely the lipids of phagocytosed blood cells. Sea-blue histiocytes arise as a result of ineffective hematopoiesis, leading to increased destruction of erythrocytes, leucocytes, and platelets and resultant saturation of normal pathways for removal of membrane lipid. Another possible mechanism causing sea-blue histiocytes to form in myelodysplastic syndromes is that the macrophages could be functionally abnormal with defective catabolic enzymes.[7]

Storage pathological condition in patients receiving long-term parenteral nutrition with fat-emulsion sources represents a further condition associated to sea-blue histiocytosis.[8]

Furthermore, infectious diseases, such as lepromatous leprosy and mononucleosis, have been also related to this condition. In the presented case, the infection by visceral leishmaniasis could be the cause of the sea-blue histiocytosis.

Thalassemia has also been related to sea-blue histiocytosis.[9]

The sea-blue histiocytes are located preferentially in the bone marrow and liver. They can also be found in the spleen, lymph nodes, lungs, and gastrointestinal tract.[10]

Accumulations of lipofuscin, glycophospholipid, and sphingomyelin within the granules become blue by May–Giemsa stain, brownish yellow on hematoxylin and eosin stain, and black on Sudam III and osmic acid.

Key Learning Points/Tips and Tricks

- The gastrointestinal tract can be affected by sea-blue histiocytosis, and this differential diagnosis should be considered.
- Careful examination of the small bowel is advised due to the bridge mucosal formation.

Scripted Voiceover

00:14 Double-balloon endoscopy by oral route revealed multiple, sessile, elevated, and confluent lesions at the duodenum.

00:59 At the jejunum, numerous sessile lesions, but also pedunculated and multilobated lesions that formed bridges throughout the lumen of the small intestine can be observed.

01:57 Due to these bridges, the balloon of the overtube was inflated proximally and only the endoscope was advanced further.

03:02 Double-balloon endoscopy by anal route demonstrated normal colonic mucosa.

03:10 At the ileocecal valve and ileum, several small sessile lesions are seen

03:28 and enhanced with indigo carmine chromoscopy.

03:34 Biopsy samples of the lesions with hematoxylin-eosin staining identified subepithelial macrophages because of the accumulation of lipofuscin, glycophospholipid, and sphingomyelin.

03:48 May-Giemsa staining revealed sea-blue macrophages, reason why this condition presents the name of sea-blue histiocytosis.

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Further Reading

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